

From gene discovery to the treatment and prevention of disease: a public health perspective

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The Human Genome Project has led to a range of predictions about the potential for new understanding of the human genome to improve medical care. While some scientists predict revolutionary advances in disease prevention and treatment, others suggest that the impact on common diseases will be limited, citing the low magnitude of risks conferred by individual genes and the complexity of gene-environment interactions. An immense gap currently exists between the scientific products of the Human Genome Project and their application for treatment and prevention of disease. Gene sequencing and discovery are only the beginning.

As outlined in the accompanying diagram, this presentation will discuss some key steps in research and development that are necessary for using genomic information to treat and prevent disease. Once a gene is suspected to play a role in disease causation, molecular research is needed to better understand gene function and epidemiological research is needed to characterize the disease risk associated with the genotype. Some basic components of genetic epidemiology include the prevalence of the genotype in populations, the magnitude of the gene-disease association, and the gene-gene and gene-environment interactions. Genetic epidemiologists will need to study not only the effects of single genes, but the effects of multiple genes interacting with each other and with environmental factors to cause disease. The Atherosclerosis Risk in Communities (ARIC) study will be discussed as an example of this type of research.

Once a gene-disease association has been well characterized through molecular and epidemiological research, the potential exists for genetic testing. Today, most genetic tests are used to diagnose patients and counsel families affected by rare, single-gene disorders. However, as associations between genes, the environment, and common diseases are discovered, more genetic tests will be proposed for newborn screening and in other public health and clinical settings. Criteria must be established for evaluating the benefits and risks of genetic testing for promoting health and preventing disease. CDC is supporting a project to develop a framework for evaluating existing data on the safety and effectiveness of DNA-based genetic tests and testing algorithms. The framework includes an assessment of the test's analytic validity, clinical validity, and clinical utility, as well as a review of the ethical, legal and social implications of testing. A case study of genetic testing for variation in *PON1* enzyme levels and pesticide sensitivity will be discussed to illustrate these concepts.

Results of genetic tests could suggest several potential avenues for disease prevention or treatment and prevention, including targeted interventions, environmental modifications, drug therapy, or perhaps gene therapy. Examples will be discussed in the context of CDC activities for integrating genomics into public health practice. As new genetic

information and technologies become available, public health agencies will have an increasing role in assessing the health needs of populations, assuring the quality of genetic tests and services, and evaluating the impact of interventions.